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NEWS 2   Jan 25   BLAST(R) searching in REGISTRY available in STN on the Web
NEWS 3   Jan 29   FSTA has been reloaded and moves to weekly updates
NEWS 4   Feb 01   DKILIT now produced by FIZ Karlsruhe and has a new update
                    frequency
NEWS 5   Feb 19   Access via Tymnet and SprintNet Eliminated Effective 3/31/02
NEWS 6   Mar 08   Gene Names now available in BIOSIS
NEWS 7   Mar 22   TOXLIT no longer available
NEWS 8   Mar 22   TRCTHERMO no longer available
NEWS 9   Mar 28   US Provisional Priorities searched with P in CA/CAplus
                    and USPATFULL
NEWS 10   Mar 28   LIPINSKI/CALC added for property searching in REGISTRY
NEWS 11   Apr 02   PAPERCHEM no longer available on STN. Use PAPERCHEM2 instead.
NEWS 12   Apr 08   "Ask CAS" for self-help around the clock
NEWS 13   Apr 09   BEILSTEIN: Reload and Implementation of a New Subject Area
NEWS 14   Apr 09   ZDB will be removed from STN
NEWS 15   Apr 19   US Patent Applications available in IFICDB, IFIPAT, and IFIUDB
NEWS 16   Apr 22   Records from IP.com available in CAPLUS, HCAPLUS, and ZCAPLUS
NEWS 17   Apr 22   BIOSIS Gene Names now available in TOXCENTER
NEWS 18   Apr 22   Federal Research in Progress (FEDRIP) now available

NEWS EXPRESS   February 1 CURRENT WINDOWS VERSION IS V6.0d,
               CURRENT MACINTOSH VERSION IS V6.0a(ENG) AND V6.0Ja(JP),
               AND CURRENT DISCOVER FILE IS DATED 05 FEBRUARY 2002
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FILE 'HOME' ENTERED AT 15:14:29 ON 08 MAY 2002

=> file .pub
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=> e perlin m/U
'U' IS NOT A VALID EXPAND FIELD CODE FOR FILE 'MEDLINE'
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SFIELDS at an arrow prompt (=>).

=> e perlin m/au
E1           4      PERLIN JONATHAN B/AU
E2          12      PERLIN L/AU
E3          56 -->  PERLIN M/AU
E4           2      PERLIN M D/AU
E5          52      PERLIN M H/AU
E6           2      PERLIN M J/AU
E7          19      PERLIN M L/AU
E8           1      PERLIN M P/AU
E9           6      PERLIN M S/AU
E10         27      PERLIN M W/AU
E11          5      PERLIN MARK/AU
E12         11      PERLIN MARK W/AU

=> e
E13         11      PERLIN MICHAEL H/AU
E14          6      PERLIN MICHAEL L/AU
E15          1      PERLIN P/AU
E16          1      PERLIN R/AU
E17          5      PERLIN R R/AU
E18         24      PERLIN S/AU
E19          8      PERLIN S A/AU
E20          8      PERLIN S I/AU
E21          4      PERLIN S J/AU
E22          5      PERLIN S M/AU
E23          2      PERLIN SUSAN A/AU
E24          2      PERLIN T/AU

=> s e3, e10, e11, e12
L1          99 ("PERLIN M"/AU OR· "PERLIN M W"/AU OR "PERLIN MARK"/AU OR "PERLIN
                MARK W"/AU)

=> s l1 and mixed
L2           2 L1 AND MIXED

=> duplicate remove l2
DUPLICATE PREFERENCE IS 'MEDLINE, BIOSIS'
KEEP DUPLICATES FROM MORE THAN ONE FILE?  Y/(N):n
PROCESSING COMPLETED FOR L2
L3           1 DUPLICATE REMOVE L2 (1 DUPLICATE REMOVED)

=> d bib ab

L3   ANSWER 1 OF 1     MEDLINE                         DUPLICATE 1
AN   2001668388     MEDLINE
DN   21571107    PubMed ID: 11714147
TI   Linear mixture analysis: a mathematical approach to resolving
     mixed DNA samples.
```

AU   **Perlin M W**; Szabady B
CS   Cybergenetics, Pittsburgh, PA 15213, USA.
AB   With the advent of PCR-based STR typing systems, **mixed** samples
        can be separated into their individual DNA profiles. Quantitative peak
        information can help in this analysis. However, despite such advances,
        forensic mixture analysis still remains a laborious art, with the high
        cost and effort often precluding timely reporting. We introduce here a new
        automated approach to resolving forensic DNA mixtures. Our linear mixture
        analysis (LMA) is a straightforward mathematical approach that can
        integrate all the quantitative PCR data into a single rapid computation.
        LMA has application to diverse mixture problems. As demonstrated here on
        laboratory STR data, LMA can assess the quality and utility of its
        solutions. Such rapid and robust methods for computer-based analysis of
        DNA mixtures may help in reducing crime.

=> s l1 and (pcr or rflp or repeat)
L4   21 L1 AND (PCR OR RFLP OR REPEAT)

=> s l1 and (pcr or rflp or snp or repeat)
L5   21 L1 AND (PCR OR RFLP OR SNP OR REPEAT)

=> duplicate remove l5
DUPLICATE PREFERENCE IS 'MEDLINE, BIOSIS'
KEEP DUPLICATES FROM MORE THAN ONE FILE? Y/(N):n
PROCESSING COMPLETED FOR L5
L6   14 DUPLICATE REMOVE L5 (7 DUPLICATES REMOVED)

=> d 1-14 bib ab

L6   ANSWER 1 OF 14   MEDLINE   DUPLICATE 1
AN   2002199470   IN-PROCESS
DN   21929801   PubMed ID: 11933190
TI   Determining sequence length or content in zero, one, and two dimensions.
AU   **Perlin Mark W**; Szabady Beata
CS   Cybergenetics, Pittsburgh, Pennsylvania.
AB   High-throughput assays are essential for the practical application of
        mutation detection in medicine and research. Moreover, such assays should
        produce informative data of high quality that have a low-error rate and a
        low cost. Unfortunately, this is not currently the case. Instead, we
        typically witness legions of people reviewing imperfect data at
        astronomical expense yielding uncertain results. To address this problem,
        for the past decade we have been developing methods that exploit the
        inherent quantitative nature of DNA experiments. By generating
        high-quality data, careful DNA-signal quantification permits robust
        analysis for determining true alleles and certainty measures. We will
        explore several assays and methods. In a one-dimensional readout, short

tandem **repeat** (STR) data display interesting artifacts. Even
with high-quality data, **PCR** artifacts such as stutter and
relative amplification can confound correct or automated scoring. However,
by appropriate mathematical analysis, these artifacts can be essentially
removed from the data. The result is fully automated data scoring, quality
assessment, and new types of DNA analysis. These approaches enable the
accurate analysis of pooled DNA samples, for both genetic and forensic
applications. On a two-dimensional surface (comprised of zero-dimensional
spots) one can perform assays of extremely high-throughput at low cost.
The question is how to determine DNA sequence length or content from
nonelectrophoretic intensity data. Here again, mathematical analysis of
highly quantitative data provides a solution. We will discuss new lab
assays that can produce data containing such information; mathematical
transformation then determines DNA length or content.
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AB   With the advent of **PCR**-based STR typing systems, mixed samples
      can be separated into their individual DNA profiles. Quantitative peak
      information can help in this analysis. However, despite such advances,
      forensic mixture analysis still remains a laborious art, with the high
      cost and effort often precluding timely reporting. We introduce here a new
      automated approach to resolving forensic DNA mixtures. Our linear mixture
      analysis (LMA) is a straightforward mathematical approach that can
      integrate all the quantitative **PCR** data into a single rapid
      computation. LMA has application to diverse mixture problems. As
      demonstrated here on laboratory STR data, LMA can assess the quality and
      utility of its solutions. Such rapid and robust methods for computer-based
      analysis of DNA mixtures may help in reducing crime.

AB   The present invention pertains to a process which can be fully automated
      for accurately determining the alleles of genetic markers. More
      specifically, the present invention is related to performing **PCR**
      amplification on locations of DNA to generate a reproducible pattern,
      labeling the **PCR** products, converting the labels into a signal,
      operating on the signal, and then determining the genotype of the location
      of the DNA. An amplification can include multiple locations from the DNA

of one or more individuals. The invention also pertains to genetics applications and systems which can effectively use this genotyping information.

L6   ANSWER 4 OF 14   MEDLINE   DUPLICATE 3
AN   1999455102   MEDLINE
DN   99455102   PubMed ID: 10523529
TI   Using quality measures to facilitate allele calling in high-throughput genotyping.
AU   Palsson B; Palsson F; **Perlin M**; Gudbjartsson H; Stefansson K; Gulcher J
CS   deCODE Genetics, Inc., 110 Reykjavik, Iceland.
SO   GENOME RESEARCH, (1999 Oct) 9 (10) 1002-12.
      Journal code: CES; 9518021. ISSN: 1088-9051.
CY   United States
DT   Journal; Article; (JOURNAL ARTICLE)
LA   English
FS   Priority Journals
EM   199912
ED   Entered STN: 20000113
      Last Updated on STN: 20000113
      Entered Medline: 19991202
AB   Currently, the main limitation in high-throughput microsatellite genotyping is the required manual editing of allele calls. Even though programs for automated allele calling have been available for several years, they have limited capability because accurate data could only be assured by manual inspection of the electropherograms for confirmation. Here we describe the development of a parametric approach to allele call quality control that eliminates much of the time required for manual editing of the data. This approach was implemented in an editing tool, Decode-GT, that works downstream of the allele calling program, TrueAllele (TA). Decode-GT reads the output data from TA, displays the underlying electropherograms for the genotypes, and sorts the allele calls into three categories: good, bad, and ambiguous. It discards the bad calls, accepts the good calls, and suggests that the user inspect the ambiguous calls, thereby reducing dependence on manual editing. For the categorization we use the following parameters: (1) the quality value for each allele call from TrueAllele; (2) the peak height of the alleles; and (3) the size of the peak shift needed to move peaks into the nearest bin. Here we report how we optimized the parameters such that the size of the ambiguous category was minimized, and both the number of miscalled genotypes in the good category and the useable genotypes in the bad category were negligible. This approach reduces the manual editing time and results in <1% miscalls.

L6   ANSWER 5 OF 14   MEDLINE   DUPLICATE 4
AN   96026116   MEDLINE
DN   96026116   PubMed ID: 7485172
TI   Toward fully automated genotyping: genotyping microsatellite markers by deconvolution.
AU   **Perlin M W**; Lancia G; Ng S K
CS   Computer Science Department, Carnegie Mellon University, Pittsburgh, PA 15213, USA.
NC   R01 NS32084 (NINDS)
SO   AMERICAN JOURNAL OF HUMAN GENETICS, (1995 Nov) 57 (5) 1199-210.
      Journal code: 3IM; 0370475. ISSN: 0002-9297.
CY   United States
DT   Journal; Article; (JOURNAL ARTICLE)
LA   English
FS   Priority Journals
EM   199511
ED   Entered STN: 19960124
      Last Updated on STN: 19960124
      Entered Medline: 19951130
AB   Dense genetic linkage maps have been constructed for the human and mouse

genomes, with average densities of 2.9 cM and 0.35 cM, respectively. These genetic maps are crucial for mapping both Mendelian and complex traits and are useful in clinical genetic diagnosis. Current maps are largely comprised of abundant, easily assayed, and highly polymorphic **PCR** -based microsatellite markers, primarily dinucleotide (CA)n **repeats**. One key limitation of these length polymorphisms is the **PCR** stutter (or slippage) artifact that introduces additional stutter bands. With two (or more) closely spaced alleles, the stutter bands overlap, and it is difficult to accurately determine the correct alleles; this stutter phenomenon has all but precluded full automation, since a human must visually inspect the allele data. We describe here novel deconvolution methods for accurate genotyping that mathematically remove **PCR** stutter artifact from microsatellite markers. These methods overcome the manual interpretation bottleneck and thereby enable full automation of genetic map construction and use. New functionalities, including the pooling of DNAs and the pooling of markers, are described that may greatly reduce the associated experimentation requirements.

AB   Advances in dinucleotide-based genetic maps open possibilities for large scale genotyping at high resolution. The current rate-limiting steps in use of these dense maps is data interpretation (allele definition), data entry, and statistical calculations. We have recently reported automated allele identification methods. Here we show that a 10-cM framework map of the human X chromosome can be analyzed on two lanes of an automated sequencer per individual (10-12 loci per lane). We use this map and analysis strategy to generate allele data for an X-linked recessive spastic paraplegia family with a known PLP mutation. We analyzed 198 genotypes in a single gel and used the data to test three methods of data analysis: manual meiotic breakpoint mapping, automated concordance analysis, and whole chromosome multipoint linkage analysis. All methods pinpointed the correct location of the gene. We propose that multipoint exclusion mapping may permit valid inflation of LOD scores using the equation max LOD - (next best LOD).

AB   Inner product mapping (IPM) has been proposed as a hybridization-based
      method for achieving low-cost, high-throughput, high-resolution radiation
      hybrid (RH) mapping of clones. Using Alu-**PCR** products of
      chromosome 11-specific clones, we serially hybridized a set of RHs against
      gridded filters of YACs having an average size of 350 kb. We then combined
      these hybridization data with preexisting RH map data to build an inner
      product map. This binning of 865 YACs provides the first high-resolution
      large-scale (> twofold redundancy) clonal coverage of human chromosome 11
      and is the first inner product map ever constructed. We verified the
      accuracy and precision of this chromosome 11 map by performing a novel
      likelihood analysis on independent YAC hybridization data. These results
      establish that IPM is a highly rapid, inexpensive, accurate, and precise
      large-scale long-range mapping method, particularly when preexisting RH
      maps are available, and that IPM can replace or complement more
      conventional short-range mapping methods. IPM may enable the rapid
      construction of sequence-ready maps and the binning of expressed
      sequences.

L6   ANSWER 8 OF 14   BIOSIS   COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN   1995:476445 BIOSIS
DN   PREV199598490745
TI   Quantitative analysis of gel electrophoresis data for automated genotyping
      applications.
AU   Richards, D. R.; **Perlin, M. W.**
CS   Computer Sci. Dep., Carnegie Mellon Univ., Pittsburgh, PA USA
SO   American Journal of Human Genetics, (1995) Vol. 57, No. 4 SUPPL., pp. A26.
      Meeting Info.: 45th Annual Meeting of the American Society of Human
      Genetics Minneapolis, Minnesota, USA October 24-28, 1995
      ISSN: 0002-9297.
DT   Conference
LA   English

L6   ANSWER 9 OF 14   MEDLINE   DUPLICATE 6
AN   95029378   MEDLINE
DN   95029378   PubMed ID: 7942856
TI   Toward fully automated genotyping: allele assignment, pedigree
      construction, phase determination, and recombination detection in Duchenne
      muscular dystrophy.
AU   **Perlin M W**; Burks M B; Hoop R C; Hoffman E P
CS   School of Computer Science, Carnegie Mellon University, Pittsburgh, PA
      15213.
NC   R01 NS32084 (NINDS)
SO   AMERICAN JOURNAL OF HUMAN GENETICS, (1994 Oct) 55 (4) 777-87.
      Journal code: 3IM; 0370475. ISSN: 0002-9297.
CY   United States
DT   Journal; Article; (JOURNAL ARTICLE)
LA   English
FS   Priority Journals
EM   199410
ED   Entered STN: 19941222
      Last Updated on STN: 20000303
      Entered Medline: 19941025
AB   Human genetic maps have made quantum leaps in the past few years, because
      of the characterization of > 2,000 CA dinucleotide **repeat** loci:
      these **PCR**-based markers offer extraordinarily high PIC, and
      within the next year their density is expected to reach intervals of a few
      centimorgans per marker. These new genetic maps open new avenues for
      disease gene research, including large-scale genotyping for both simple
      and complex disease loci. However, the allele patterns of many
      dinucleotide **repeat** loci can be complex and difficult to
      interpret, with genotyping errors a recognized problem. Furthermore, the
      possibility of genotyping individuals at hundreds or thousands of

polymorphic loci requires improvements in data handling and analysis. The automation of genotyping and analysis of computer-derived haplotypes would remove many of the barriers preventing optimal use of dense and informative dinucleotide genetic maps. Toward this end, we have automated the allele identification, genotyping, phase determinations, and inheritance consistency checks generated by four CA **repeats** within the 2.5-Mbp, 10-cM X-linked dystrophin gene, using fluorescein-labeled multiplexed **PCR** products analyzed on automated sequencers. The described algorithms can deconvolute and resolve closely spaced alleles, despite interfering stutter noise; set phase in females; propagate the phase through the family; and identify recombination events. We show the implementation of these algorithms for the completely automated interpretation of allele data and risk assessment for five Duchenne/Becker muscular dystrophy families. The described approach can be scaled up to perform genome-based analyses with hundreds or thousands of CA-**repeat** loci, using multiple fluorophors on automated sequencers.

L6   ANSWER 10 OF 14   MEDLINE
AN   96039037   MEDLINE
DN   96039037   PubMed ID: 7584409
TI   Intelligent DNA-based molecular diagnostics using linked genetic markers.
AU   Pathak D K; Hoffman E P; **Perlin M W**
CS   Department of Computer Science, Carnegie Mellon University, USA.
SO   ISMB, (1994) 2 331-9.
     Journal code: CCP; 9509125.
CY   United States
DT   Journal; Article; (JOURNAL ARTICLE)
LA   English
FS   Priority Journals
EM   199512
ED   Entered STN: 19960124
     Last Updated on STN: 19960124
     Entered Medline: 19951214
AB   This paper describes a knowledge-based system for molecular diagnostics, and its application to fully automated diagnosis of X-linked genetic disorders. Molecular diagnostic information is used in clinical practice for determining genetic risks, such as carrier determination and prenatal diagnosis. Initially, blood samples are obtained from related individuals, and **PCR** amplification is performed. Linkage-based molecular diagnosis then entails three data analysis steps. First, for every individual, the alleles (i.e., DNA composition) are determined at specified chromosomal locations. Second, the flow of genetic material among the individuals is established. Third, the probability that a given individual is either a carrier of the disease or affected by the disease is determined. The current practice is to perform each of these three steps manually, which is costly, time consuming, labor-intensive, and error-prone. As such, the knowledge-intensive data analysis and interpretation supersede the actual experimentation effort as the major bottleneck in molecular diagnostics. By examining the human problem solving for the task, we have designed and implemented a prototype knowledge-based system capable of fully automating linkage-based molecular diagnostics in X-linked genetic disorders, including Duchenne Muscular Dystrophy (DMD). Our system uses knowledge-based interpretation of gel electrophoresis images to determine individual DNA marker labels, a constraint satisfaction search for consistent genetic flow among individuals, and a blackboard-style problem solver for risk assessment. We describe the system's successful diagnosis of DMD carrier and affected individuals from raw clinical data.

L6   ANSWER 11 OF 14 BIOSIS   COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN   1994:525359 BIOSIS
DN   PREV199497538359
TI   Automated genotyping of dinucleotide **repeat** markers.
AU   **Perlin, M. W. (1)**; Hoffman, E. P.

```
CS    (1) Computer Sci., Carnegie Mellon Univ., Pittsburgh, PA USA
SO    American Journal of Human Genetics, (1994) Vol. 55, No. 3 SUPPL., pp.
      A199.
      Meeting Info.: 44th Annual Meeting of the American Society of Human
      Genetics Montreal, Quebec, Canada October 18-22, 1994
      ISSN: 0002-9297.
DT    Conference
LA    English


L6    ANSWER 12 OF 14  BIOSIS  COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN    1993:444537  BIOSIS
DN    PREV199345080162
TI    Detection of indicator bacteria and pathogens in water by polymerase chain
      reaction (PCR) and gene probe methods.
AU    Atlas, R. M. (1); Bej, A. (1); Mahbubani, M. (1); Steffan, R. (1);
      Perlin, M. (1); Dicesare, J.; Haff, L.
CS    (1) Dep. Biol., Univ. Louisville, Louisville, KY USA
SO    Mongkolsuk, S. [Editor]; Lovett, P. S. [Editor]; Trempy, J. E. [Editor].
      (1992) pp. 219-223. Biotechnology and environmental science: Molecular
      approaches.
      Publisher: Plenum Press 233 Spring Street, New York, New York, USA.
      Meeting Info.: International Conference Bangkok, Thailand August 21-24,
      1990
      ISBN: 0-306-44352-X.
DT    Article
LA    English


L6    ANSWER 13 OF 14      MEDLINE                        DUPLICATE 7
AN    92152848      MEDLINE
DN    92152848    PubMed ID: 1785923
TI    Detection of Giardia cysts by using the polymerase chain reaction and
      distinguishing live from dead cysts.
AU    Mahbubani M H; Bej A K; Perlin M; Schaefer F W 3rd; Jakubowski
      W; Atlas R M
CS    Department of Biology, University of Louisville, Kentucky 40292.
SO    APPLIED AND ENVIRONMENTAL MICROBIOLOGY, (1991 Dec) 57 (12) 3456-61.
      Journal code: 6K6; 7605801. ISSN: 0099-2240.
CY    United States
DT    Journal; Article; (JOURNAL ARTICLE)
LA    English
FS    Priority Journals
EM    199203
ED    Entered STN: 19920405
      Last Updated on STN: 19920405
      Entered Medline: 19920316
AB    A method was developed for the detection of Giardia cysts by using the
      polymerase chain reaction (PCR) and the giardin gene as the
      target. DNA amplification by PCR, using giardin DNA as the
      target, resulted in detection of both live and dead cysts. When giardin
      mRNA was used as the target, the ability to amplify cDNA by PCR
      depended on the mode of killing. Cysts killed by freezing were not
      detected by PCR when giardin mRNA was the target. Cysts killed
      by heating or exposure to monochloramine, however, gave positive detection
      signals for both DNA and giardin mRNA targets. The amount of giardin mRNA
      and total RNA was significantly increased in live cysts following the
      induction of excystation. Cysts killed by freezing, heating, or exposure
      to monochloramine did not show a change in RNA content. The detection of
      the giardin gene by PCR permits a sensitive and specific
      diagnosis for Giardia spp. Discrimination between live and dead cysts can
      be made by measuring the amounts of RNA or PCR-amplified product
      from the giardin mRNA target before and after the induction of
      excystation.


L6    ANSWER 14 OF 14  BIOSIS  COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN    1991:381182  BIOSIS
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DN   BR41:53572
TI   DETECTION OF VIABLE LEGIONELLA AND GIARDIA USING **PCR** AND GENE
     PROBES.
AU   MAHBUBANI M; BEJ A; **PERLIN M**; SCHAEFER F; ATLAS R M
CS   UNIV. LOUISVILLE, LOUISVILLE, KY.
SO   91ST GENERAL MEETING OF THE AMERICAN SOCIETY FOR MICROBIOLOGY 1991,
     DALLAS, TEXAS, USA, MAY 5-9, 1991. ABSTR GEN MEET AM SOC MICROBIOL. (1991)
     91 (0), 87.
     CODEN: AGMME8.
DT   Conference
FS   BR; OLD
LA   English

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COST IN U.S. DOLLARS   SINCE FILE   TOTAL
                                                 ENTRY   SESSION
FULL ESTIMATED COST   17.46   17.67

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